

MITOCHONDRIAL DNA AUTOSCORING SYSTEM
Stockwell et al.
 Appl. No.: Unknown Atty Docket: ABIOS.042A

REPLACEMENT SHEET

System components and tunable parameters

Program	Function performed
BlastParse.pl	Parse BLAST output
mark_substitution_heteroplasmy.pl	Mark ambiguous base calls
extract_SE_consensus.pl	Extract a consensus sequence from a phrap polyphred run, replacing low quality bases with '?', and polyphred rank 1 bases with 'N'.
compute_coverage.pl	Calculates overall, forward strand, and reverse strand coverage for all bases in the consensus sequence
count_hv1_deletes.pl	Count deletions in HV1 relative to the rCRS
border_index.pl	Compute start and end positions of HV1 and HV2 regions.
fix_mitotype_reporting_range.pl	Output a mtDNA profile based on input variants list and reportable range.
find_bad_traces_from_blast_report.pl	Reject reads that do not align appropriately to the rCRS
determineReadTypes.pl	Adds template name, template type, and primer type to phred output files.
seq2delta_vs	Align mtDNA profile to rCRS and report variants according to nomenclature.
calculate_coverage_mitotype.pl	Mask mtDNA consensus sequence based on required coverage and calculated coverage
count_hv1_inserts.pl	Count insertions in HV1 relative to the rCRS
generate_hv_mask_fasta_files.pl	Extract the HV1 portion and HV2 portion of the mtDNA consensus sequence, based on the computed HV1 and HV2 regions.
flip_fasta.pl	Invert an X masked sequence output by cross match

FIG. 5

HONOLULU DNA AUTOSCUENING SYSTEM REPLACEMENT SHEET
 Stockwell et al.
 Appl. No.: Unknown Atty Docket: ABIOS.042A

	Assessment 1	Assessment 2
Total True Positives: $N_{\text{automated}} = N_{\text{manual}}$	16	10
Total True Negatives: $A_{\text{automated}} = A_{\text{manual}}$	77,358	110,354
Total False Positives: $N_{\text{automated}} = A_{\text{manual}}$	95	807
Total False Negatives: $A_{\text{automated}} = N_{\text{manual}}$	6 ^a	14
Total Incorrect	0	0
Sensitivity: $TP/(TP+FN)$	72.73%	41.67%
Specificity: $TN/(TN+FP)$	99.88%	99.27%
Positive Predictive Value: $TP/(TP+FP)$	14.41%	1.22%
Negative Predictive Value: $TN/(TN+FN)$	99.99%	99.99%

^a Predominate base correctly called
 Differences take into account the consensus of the two analysts

FIG. 6